



**INTERNATIONAL  
RARE DISEASES RESEARCH  
CONSORTIUM**

**Minutes of the 11<sup>th</sup> Executive  
Committee meeting**

2 October 2014



**IRDIRC**

## EXECUTIVE SUMMARY

The Executive Committee (Exec Comm) of the International Rare Diseases Research Consortium (IRDiRC) met on 2 October 2014 by teleconference. The eleventh meeting of the Exec Comm brought together 28 participants, including Exec Comm members and representatives of two Scientific Committees (Sci Comm).

Two new members were introduced: Dr Denis Lacombe representing the European Organisation for Research and Treatment of Cancer, and Dr Mao Mao from WuXi AppTec.

The Chair of the Exec Comm reported his recent activities related to recruitment for IRDiRC and representation of IRDiRC at conferences. It was suggested that a member of the Rare and Undiagnosed Diseases group be invited to the Diagnostics Sci Comm to link the two initiatives. With regard to the possibility to organize an IRDiRC conference in the United Arab Emirates, Exec Comm agreed that:

- ▶ IRDiRC conferences should be organized in Europe or the US to favor participation of the members of the Sci Comm and Working Groups (WG).
- ▶ For scientific outreach conference, IRDiRC should cooperate with organizers to develop the scientific program, without responsibility for practical and financial aspects of the conference organization.

The nomination of two new members to the Sci Comm was approved by the Exec Comm for a 3-year mandate. They are: Dr Feng Zhang (nominated by WuXi AppTec) for the Diagnostic Sci Comm and Virginie Hivert (nominated by Eurordis) for the Therapies Sci Comm.

The Exec Comm continued discussion on possible mechanisms to fund proposals for tools presented by the Sci Comms. Members reported progress in this direction such as allocation of fund, modification of internal rules for funding, possibility to fund workshops, etc. However, more visibility on these projects was requested, and the process for selection and funding still needs to be defined. This topic will be further discussed at the next face-to-face meeting of the Exec Comm in Shenzhen, China, in November.

The Exec Comm reviewed and commented on the document specifying the definition, criteria and procedure for proposals to obtain the “IRDiRC Recommended” label. The document needs further revision by the Sci Comms, together with the Scientific Secretariat, before adoption by the Exec Comm.

The Scientific Secretariat provided an update on the organization of the 2<sup>nd</sup> IRDiRC conference, Shenzhen, China, and the organization of IRDiRC sessions at international conferences.

The Exec Comm also agreed on:

- ▶ The election of a vice-chair of the Exec Comm, electronically, in November.
- ▶ The organization of the next face-to-face meetings of the Exec Comm in February/March 2015 in Europe and in September/October 2015 in the US, coupled to other conferences.
- ▶ The delay of two weeks for the members of the Exec Comm to express any concerns about the transition of IRDiRC as a member organization of the Global Alliance for Genomics and Health.

## REPORT

### Presentation of new members

#### European Organisation for Research and Treatment of Cancer (EORTC)

The EORTC, a not-for-profit research organization conducting pan-European cancer clinical trials, with a focus in rare tumors, was presented. Through its history and its international reach - 25 countries are involved - EORTC has developed a large database of patients. Capacity of recruitment is between 3,000-7,000 patients on a yearly basis, all cancers concerned. At any time, EORTC manages a volume of about 20 trials with a turnover of 8-10 trials per year. EORTC is also working on the molecular biology of rare cancers to accumulate biological knowledge. All operations are coordinated by the 170 staff members from the headquarters in Brussels.

#### WuXi AppTec

WuXi AppTec, a research and development services company with operations in both US and China, employing 8,000 people in 12 sites, was presented. WuXi AppTec provides a broad and integrated portfolio of services throughout the drug R&D process for pharmaceutical companies, including a genome center and large scale manufacturing.

### Report from the Chair

#### Outcomes of the International Conference on Rare and Undiagnosed Diseases, Rome, Italy, 29-30 September 2014

The purpose of this conference, organized by the NIH National Center for Advancing Translational Sciences (NCATS), was to discuss data sharing and collaboration among projects internationally that are aiming to diagnose patients with ultra rare diseases.

It would be beneficial for IRDiRC to work in concert with this group. A link could be created by inviting a member of this group to join the Diagnostics Sci Comm, either through nomination by one of the NIH Institutes that has not yet nominate a candidate or by the Rare and Undiagnosed Diseases group.

## Discussions with organizations in Saudi Arabia and the UAE interested in joining IRDiRC

The Chair had had discussions with organizations from Saudi Arabia and United Arab Emirates (UAE).

- ▶ Saudi Human Genome Project submitted a letter of intent earlier this week. *Post-meeting note: the application was accepted on 3 October 2014.*
- ▶ Paul Lasko will visit institutions in Dubai and Sharjah in the UAE in February 2015.

The Chair also informed the Exec Comm that organizations in the UAE are interested in hosting an IRDiRC conference, and invited the Exec Comm to consider this proposition.

## IRDiRC and scientific outreach conferences

The Chair suggested the possibility to develop two kinds of conference:

- ▶ IRDiRC conference: organized by IRDiRC with full responsibility.
- ▶ Scientific outreach conference: IRDiRC would cooperate with organizers to develop the scientific program, without responsibility for practical and financial aspects of the conference organization.

Members of the Exec Comm agreed that the two types of conference would be beneficial:

- ▶ IRDiRC conferences are a good opportunities for members of the Sci Comms and WGs to meet and network. Such conference should be organized either in Europe or the US in the future to limit travel expenses. Financial support of such conferences is however a concern and thus annual meetings are not possible.
- ▶ Outreach activities constitute an important role of IRDiRC to, among others, help countries to develop a rare disease strategy by providing context, network and support. Sci Comms could be involved in the preparation of the scientific program as they did for the Dublin and Shenzhen IRDiRC conferences.

⇒ This topic will be further considered later, but this preliminary discussion did bring the Chair useful information on how to conduct his meeting in Dubai.

## Discussion on the principle to elect a vice-chair for the Executive Committee

The unavailability of the Chair to participate in the next Exec Comm meeting in November in Shenzhen brought to light the fact that there is no procedure for the replacement of the Chair when necessary. The proposition to elect a vice-chair was approved by the Exec Comm.

It was decided that:

- ▶ Nominations should be sent to the Scientific Secretariat before the Shenzhen Executive Committee meeting. *Post-meeting note: please submit by 31 October 2014.*
- ▶ Election of the vice-chair will be conducted by email as all the members of the Executive Committee will not be present in Shenzhen.
- ▶ Vice-chair should consider becoming the next Chair by participating to the election that will be open to other candidates.

The question of the possibility of two terms for the chair was raised. *Notes post-meeting: the IRDiRC governance states that “Chair is elected for a maximum period of three years.”*

### **Nominations for the Scientific Committees**

The nomination of two new members of the Sci Comm was approved by the Exec Comm for a 3-year mandate. The new members are:

#### Diagnostics Sci Comm:

- ▶ Dr Feng Zhang (nomination by WuXiAppTec)

#### Therapies Sci Comm:

- ▶ Dr Virginie Hivert (nomination by Eurordis, to replace Maria Mavris)

### **Options to select and fund short-term projects directly supporting IRDiRC activities**

Diagnostics and Interdisciplinary Sci Comms presented a total of four short-term projects to be funded in the 10<sup>th</sup> meeting of the Exec Comm (June 2014). Funding of such projects is challenging as there is no adapted process to coordinate international funding from several types of funders with different constraints.

However, members of the Exec Comm reported some progress:

- ▶ Genome Canada and CIHR have committed funds for a Canadian consortium that will support international activities for Global Alliance for Genomics and Health. As there is an overlapping mandate between IRDiRC and this organization, this fund could support the Canadian contribution to the projects proposed within IRDiRC, through an international peer review process.
- ▶ AFM-Telethon has internally evaluated the best way to participate in an international co-funding of such projects. The process is being finalized and will be put through for the approval of the board.

- ▶ E-Rare 3, which is unable to fund projects outside official calls, plans to work closely with IRDiRC Sci Comms to design the future calls and their topics. A closer connection between E-Rare, IRDiRC and its Sci Comms will be helpful in the future development of funding models.
- ▶ The European Commission (EC) can support in many other ways than launching calls for proposal. For example, the EC would be able to fund meeting/workshop for action point of the Sci Comms. In addition, the EC is starting to work on multiannual programs.

These discussions indicate several expression of interest and a beginning of reaction to support the proposals from Sci Comms that were unexpected from the start and necessitate creativity.

However, there is a necessity:

- ▶ To improve visibility about these projects (advancement, funding, etc.) to facilitate decision-making by each interested members.
  - ▶ To review the document presenting options for mechanisms for selection and funding of short-term projects proposed by the Sci Comms for decision.
- ⇒ This topic will be an expanded agenda item at the next face-to-face meeting of the Exec Comm in Shenzhen in November.
- ⇒ The Exec Comm would like an update from the Sci Comms about the progress of the proposed projects at the Shenzhen meeting.

### “IRDiRC Recommended” procedure activities

Although the process for adoption described below was already agreed on, further definition of the process is necessary to extend the basic mechanism:

- ▶ Sci Comm to produce 1-2 pages of rationale together with list of other most important tools or standards, and contentious issues.
- ▶ Consultation with the two other Sci Comms.
- ▶ Submission to the Exec Comm for approval.

Exec Comm commented the document prepared by the Scientific Secretariat proposing further definition, criteria and procedure:

- ▶ The last sentence of the “Proposed definition” should be changed to specify that “IRDiRC will encourage/expect some sustainability plan for these resources” as the terminology “IRDiRC **supports** the long-term sustainability of the resources” is not considered as acceptable.
- ▶ Review of the proposals will be conducted in rolling process, not annually as suggested.

- ▶ Process for application and review will be established to allow applications from outside IRDiRC Sci Comms/WGs.
  - ▶ The document is not clear on what type of entity could receive that designation as the terminology is not consistent throughout the document: tool/ standard/ guideline/ service.
  - ▶ Considering the lack of expertise of the Exec Comm for the technical part of the proposals, it was suggested that the Sci Comms/WGs will review the proposals and provide an opinion to the Exec Comm that will rule if the judgement is based on scientific ground or if there is any conflict of interest, acting as an advisory council.
  - ▶ Other points to consider:
    - Maintenance of these recommendations: should a periodic review be implemented?
    - Justification if the decision for recommendation is negative
- ⇒ The Scientific Secretariat will revise the document - process for application, how to evaluate, length of recommendation, etc. – to be reviewed by the Sci Comms before adoption by the Exec Comm.

### **Agenda of the face-to-face meeting in Shenzhen**

Members of the Exec Comm agreed that the following topics should be included in the agenda of the Shenzhen Exec Comm meeting:

- ▶ Roundtable with “State-of-play” of funding initiatives
- ▶ State of play of rare diseases research in China
- ▶ “IRDiRC Recommended” procedure
- ▶ Selection and funding of proposals from the Sci Comm
- ▶ Review of gap analysis based on a summary report of Sci Comms and WGs analysis, and a summary report on trends in rare disease research based on the literature
- ▶ Scheduling of next 2-3 Exec Comm meetings
- ▶ Decision on the transition to Global Alliance for Genomics and Health organizational membership

### **Proposal to have an Executive Committee meeting in Europe in January/February**

Members of the Exec Comm expressed their preference to have the next face-to-face meeting of the Exec Comm in February/March, preferably associated with another conference.

The Scientific Secretariat will research international conferences around February/March 2014 in Europe, and September/October 2014 in the US, to couple the Exec Comm meetings with other conferences/meetings.

### **Update on IRDiRC conferences**

#### **2nd IRDiRC conference in Shenzhen**

- ▶ The final draft of scientific program is under review by the Scientific Committee.
- ▶ By 2 October, around 75 foreign participants were registered - mostly from Europe, from both industry and academia. Number of Chinese participants, who tend to register at the last minute, is difficult to estimate.
- ▶ The deadline for poster abstract submission was extended to 15 October 2014. Around 15 abstracts had been reviewed and accepted by the Scientific Committee. A couple more abstracts have been received since the extension of the deadline and will be reviewed shortly.

#### **Organization of IRDiRC session at international meetings**

The Scientific Secretariat is now organizing IRDiRC session at international conference. The next session will be held on 9 October 2014 at the IX annual meeting of ICORD, Ede, Netherlands, with the following talks:

- ▶ Recommendations to speed up the R&D process in the field of orphan drugs.
- ▶ Initiatives to speed up data mining in the field of rare diseases.
- ▶ Initiatives to speed up data sharing.

Other conferences suggested by either Exec Comm members or the Scientific Secretariat:

- ▶ BIO conference
- ▶ DIA conference
- ▶ European Society of Human Genetic conference
- ▶ American Society Human Genetic conference
- ▶ World Orphan Drug Congress
- ▶ Human Genetic Society of Australasia

The conferences should be chosen in function of the target. Some members do not consider organizing IRDiRC sessions at DIA and World Orphan Drug Congress as suitable whereas other members regard these conferences to be appropriate to reach potential industry members.

⇒ Exec Comm members are invited to send suggestions to the Scientific Secretariat.

### **Membership for Global Alliance for Genomics and Health**

IRDiRC is a signatory of the non-binding Letter of Intent for Global Alliance for Genomics and Health and thus considered as a partner organization. With the publication of their constitution, the Global Alliance is proposing, to its partners, a transition to member organizations.

As general information:

- ▶ Individuals from Organizational Members may play various roles, including membership of Working Groups, other committees, and the Steering Committee. Each Organizational Member will have the right to designate individuals from its organization who will represent the organization in Global Alliance-related work, meetings, and governance matters.
- ▶ The membership agreement is not intended to and does not create any legally binding financial or other obligation on the part of the signatory organization

Although strategically, it is better to become member to have influence and combine force with them, some members of the Exec Comm expressed their reservations as the governance of the Global Alliance is not yet well defined - rights and responsibility of member/partner, representation in committee, process for decision - and that more transparency would be beneficial to decision-making.

⇒ Members are given two weeks to consider the proposition of transition. Any member disagreeing with the transition should inform the Chair of the Exec Com. Transition will only be accepted if absolute consensus is reached.

**Annex - List of participants**

| <b><u>Members</u></b>   | <b><u>Representative</u></b>                   |
|---|--|
| IRDiRC Chair Executive Committee,<br>Canadian Institutes of Health Research,<br>Canada  | Paul Lasko                                     |
| Western Australian Department of<br>Health, Australia                                   | Hugh Dawkins                                   |
| EORTC-European Organisation for<br>Treatment & Research on Cancer,<br>Belgium           | Denis Lacombe                                  |
| Chinese Rare Disease Research<br>Consortium, China                                      | Qing Wang                                      |
| WuXi AppTec, China  | Mao Mao  |
| E-RARE-2 (E-Rare Group of Funders),<br>Europe   | Daria Julkowska                                |
| European Commission, (DG Research<br>and Innovation), EU                                | Anders Colver, Iiro Eerola and Irene Nordstedt |
| EURORDIS ( Patient Advocacy Group),<br>Europe   | Béatrice de Montleau                           |
| AFM- French Association against<br>Myopathies, France                                   | Françoise Rouault                              |
| ANR- French National Research<br>Agency, France   | Daria Julkowska                                |
| FFRD – French Foundation for rare<br>diseases, France                                   | Nicolas Lévy                                   |
| Lysogene, France  | Karen Aiach                                    |
| Children’s New Hospitals Management<br>Group, Georgia                                   | Oleg Kvlivdize                                 |
| Shire, Ireland  | Emmanuel Chantelot                             |
| Telethon Foundation, Italy  | Lucia Monaco                                   |
| The Netherlands Organisation for<br>Health Research and Development,<br>The Netherlands | Sonja van Weely                                |
| Carlos III Health Institute, Spain  | Pedro Cortegoso Fernandez                      |
| Food and Drug Administration, USA   | James Reese                                    |
| Genetic Alliance, USA   | Sharon Terry                                   |
| National Eye Institute, NIH, USA  | Santa Tumminia                                 |
| National Human Genome Research<br>Institute, NIH, USA                                   | Jeff Schloss                                   |
| NORD, USA   | Peter Saltonstall                              |
| PTC Therapeutics, USA   | Diane Goetz                                    |
| Sanford Research, USA   | David Pearce                                   |
| <b><u>Scientific Committees</u></b>   |  |
| Chair Diagnostic Sci Com  | Kym Boycott                                    |
| Chair Therapies Sci Com   | Yann Le Cam                                    |

|                                      |  |
|--------------------------------------|--|
| <b>IRDIRC Scientific Secretariat</b> |  |
| SUPPORT-IRDiRC project               | Barbara Cagniard, Lilian Lau and Sophie Höhn |

## Apologies

| <b><u>Members</u></b>   | <b><u>Representative</u></b> |
|---|------------------------------|
| Genome Canada, Canada   | Pierre Meulien               |
| BGI, China  | Ning Li                      |
| European Commission, (DG Health and Consumer Protection), EU                    | Stefan Schreck               |
| Academy of Finland, Finland   | Heikki Vilen                 |
| Federal Ministry of Education and Research, Germany                             | Ralph Schuster               |
| Chiesi Farmaceutici, Italy  | Andrea Chiesi                |
| Istituto Superiore de Sanita, Italy   | Enrico Garaci                |
| Korea National Institute of Health, Korea                                       | Hyun-Young Park              |
| Prosensa, The Netherlands   | Luc Dochez                   |
| National Institute for Health Research, United Kingdom                          | Willem Ouwehand              |
| Genzyme, USA  | Carlo Incerti                |
| Isis Pharmaceuticals, USA   | Brett Monia                  |
| National Cancer Institute, NIH, USA   | Edward Trimble               |
| National Center for Advancing Translational Sciences, NIH, USA                  | Christopher Austin           |
| National Institute of Arthritis and Musculoskeletal and Skin Diseases, NIH, USA | Stephen Katz                 |
| National Institute of Child Health and Human Development, NIH, USA              | Melissa Parisi               |
| National Institute of Neurological Disorders and Stroke, NIH, USA               | Danilo Tagle                 |
| NKT Therapeutics, USA   | Robert Mashal                |
| Office of Rare Diseases, USA  | Pamela McInnes               |
| <b><u>Scientific Committees</u></b>   |                              |
| Chair Interdisciplinary Sci Com   | Hanns Lochmüller             |



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